

Quantifying Variation 6mins

Sunday, 25 October 2015 7:03 PM

- Allele, genotypic or phenotypic frequency

Allele Frequency

$F(\text{Allele}) = (\# \text{ of Homozygotes for the allele} \times 2 + \# \text{ of Heterozygotes for the allele}) / \text{Total number of alleles}$

$$\sum F(\text{Allele}) = 1$$

Broad Sense

- Denoted by H^2
- Inclusion of all potential sources of genetic variation such as dominance, epistasis, maternal and paternal effects
- $H^2 = V_{\text{genotype}} / V_{\text{phenotypic}}$

Narrow Sense

- Denoted by h^2
- Proportion of total phenotypic variation that is due to the additive effects of genes
- $h^2 = V_{\text{additive}} / V_{\text{phenotypic}}$

Hardy Weinberg Equilibrium

- HW means constant genotypic frequency due to constant allele frequency
- HW is a calculation of expected genotypic frequencies
- $P^2 + 2pq + q^2 = 1$
 - o P and q are allele frequencies
 - o $P^2 = f(\text{AA})$
 - o $2pq = f(\text{Aa})$
 - o $Q^2 = f(\text{aa})$
- Dominance does not mean increasing frequency
- Constant genotypic frequency does not mean HW but HW means constant genotypic frequency
- Assumptions:
 - o Random Mating
 - o No Selection
 - o No Migration or gene flow
 - o No Mutation
 - o Infinite population: large enough to prevent sampling errors and random effects
- One round of HW does not bring back HW equilibrium
- Populations that are not evolving have genotypic frequencies that do not change across generations

χ^2 - Chi Square

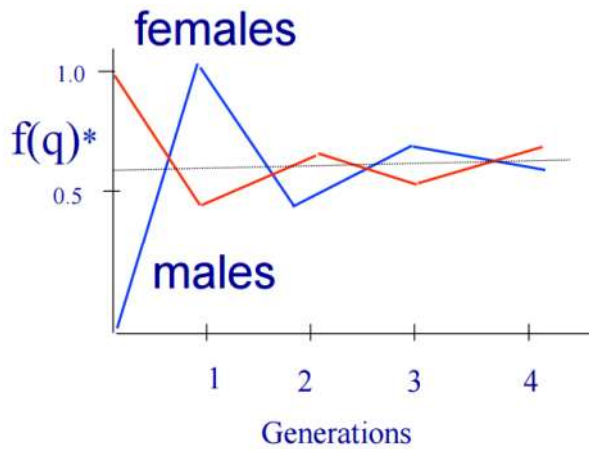
- $\chi^2 = \sum (O-E)^2/E$
- H_0 : Observed difference/error is due to chance
- $p < 0.05$ then we reject H_0

Heterozygote Undetermined with Complete Dominance

- Allele frequency of the recessive allele is the square root of the corresponding phenotype assuming HW
- $Q^2 = f(\text{aa})$
- $f(a) = \sqrt{q^2}$
- Less accurate method
- Homozygote and heterozygote's must be distinguishable

X-Linked

- Allele frequency is the frequency of the trait in males
- If HW the allele frequency is also the same in females
- Cannot restore HW after nonHW in one round of HW as it takes multiple HW generations



Selection

- Fitness measure: Proportion of mature fertile offspring left by an individual
- Relative fitness (w)
 - o 0 if no offspring is left or if they are infertile
 - o 1 for genotype that leaves the most offspring
- Selection coefficient (s)
 - o $W = 1-s$
 - o $S = 1$, animal is dead or there is no offspring

$F(G_1) = f(G_0)w$ /proportion of offspring going onto G_1

G_0 - HbA = 0.8 HbS = 0.2 (allele frequency)

Assume H-W at G_0	(G_0)	Relative fitness	(G_1)	Genotypic Frequency in G_1
$f(\text{HbAHbA})$	0.64 $p^2 = (0.8 \times 0.8)$	1	$0.64 \times 1 = 0.64$ 0.96	0.67
$f(\text{HbAHbS})$	0.32 $2pq = 2 \times 0.8 \times 0.2$	1	$0.32 \times 1 = 0.32$ 0.96	0.33
$f(\text{HbSHbS})$	0.04 $q^2 = 0.2 \times 0.2$	0	$0.04 \times 0 = 0$	

0.96 = 0.64 + 0.32 the proportion going on to next generation

$$F(A) = [F(G_1)_{\text{homozygote}} \times 2 + F(G_1)_{\text{heterozygote}}] / 2$$

Selective Agents

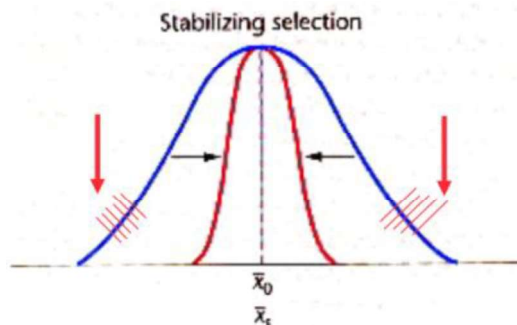
- Climate
- Pollution
- Predators
- Disease
- Insecticide
- Antibiotics

Stable Polymorphism

- Heterozygote form is the fittest
- Eg. Sickle cell in locations with malaria
- Equilibrium due to selection not HW

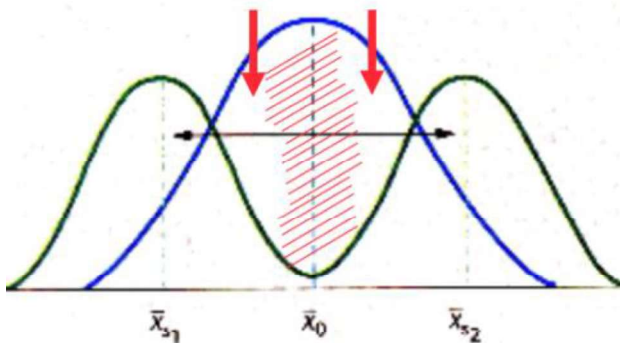
Stabilisation selection

- Selection against extremes in phenotype



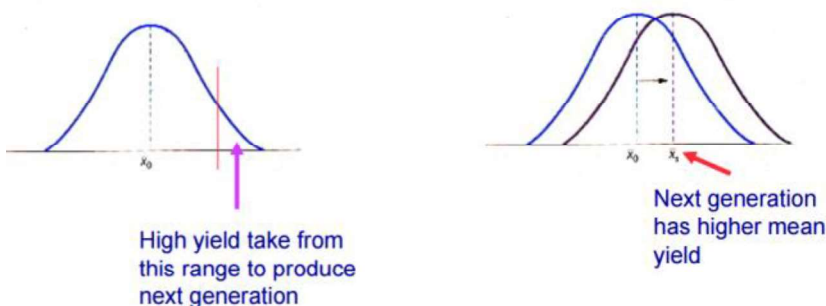
Disruptive Selection

- Selection against the intermediate, often common phenotypes



Directional Selection

- Selection for one phenotype from the end of the phenotype and presumably genotype range
- Artificial selection is usually directional selection



Mutation

- On its own it has a minor change to allele frequency
- Source of all new alleles
- Can have a significant effect on allele frequency if selection also occurs

Migration

- $\Delta p = m(x-p)$
 - o Δp = change in p , $f(a)$ in one generation
 - o M = migration rate, proportion of migrant genes entering resident population per generation
- $1-m$ = proportion of resident genes after one generation
- $X = f(A)$ of migrants
- $P = f(A)$ of residents
- $f(A)_{total} = p(1-m)+mx$
- If allele frequency is the same, $p=x$ then migration has no effect

Non-random Mating

- Genetic relation: Mating between related individuals such as inbreeding, consanguinity, self fertilisation
- Assortative Mating (AM): Not genetically related
 - o Positive Assortative Mating:
 - Select mates with similar traits
 - Humans do this
 - o Negative Assortative Mating:
 - Select mates with opposite/different traits
 - Increase heterozygosity sometimes if phenotype has a genetic cause
- One individual contributes to a large number of offspring

Self-fertilisation

- Allele frequency the same
- Eventually only homozygote genotypes remain which changes the genotypic frequency

Consanguinity

- Alters genotypic ratios by increasing homozygotes
- Allele frequency remains the same

Relations

- 1st Cousin once removed: A parents cousin
- 2nd Cousin: Same great-grandparents
- Double 1st Cousin: Offspring of two different 1st Cousins

Population Size

- Population size needs to be large enough so change is not significant nor changes the allele or genotypic frequency.

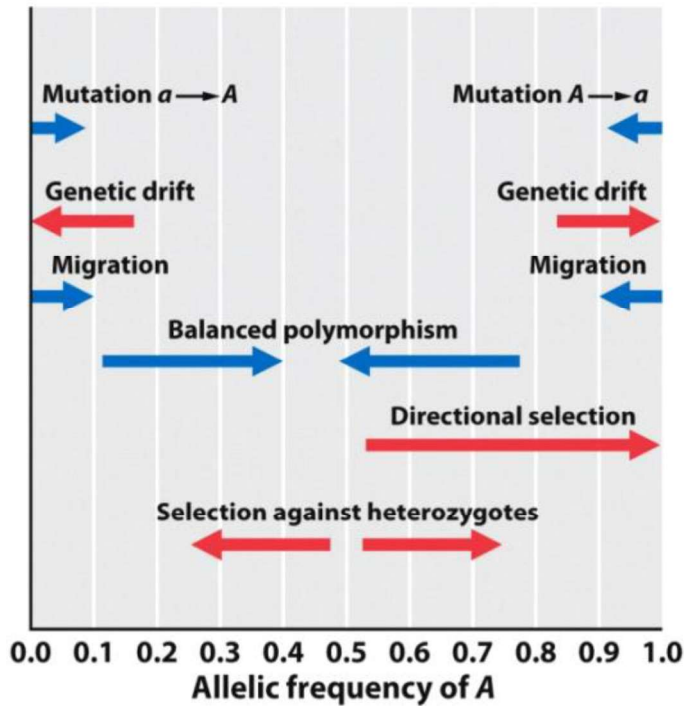
Genetic Drift

- Random change in allele frequency due to sampling error
- Occurs in all populations but effect pronounced in small populations
- Founder effect: Small group left larger group with an allele which increases frequency within that group
- Bottleneck: Population goes through a severe reduction and only a few survive and reproduce

Modern Synthetic Theory of Evolution

1. Genetic Variability
 - a. Gene mutation
 - b. Change in chromosome
 - c. Structure and number
 - d. Genetic recombination
 - e. Gene flow
2. Phenotypic Variation
3. Allele frequency different between populations
 - a. Evolutionary Forces
4. Speciation

a. Reproductive isolation



Species

- Members of a species share a common gene pool

Speciation

- Separation of a gene pool
- Forces pushing allele frequency apart if greater than the forces keeping the allele frequencies the same
- Allopathic: Physical barrier splitting populations
- Parapatric: Similar to allopathic but occurs in adjacent regions
- Sympathic: Separation of gene pool within same area
 - o Eg. Chromosome number imbalance

Polyploidy

- Organisms have more than two sets of chromosomes which is common in plants
- AA and BB form a AB hybrid which is sterile

Population isolation

- Habitat/geological/Ecological: Occupy different habitats
- Seasonal/temporal: Different timing of reproduction
- Ethological: Behaviour, often a ritual before mating
- Mechanical: Physically unable to fertilise possible due to pollinator restrictions
- Physiological: Gametes make it to female but do not survive
 - o Only certain pollen form a pollen tube
 - o Spermicide in vagina
- Gametic isolation: Inviability of male gametes in female reproductive tract

Post-zygote mechanism

- Fertilisation occurs and a hybrid zygote is formed
 1. Hybrid inviability or breakdown: Dies after birth
 2. Development of hybrid sterility
 3. F2 breakdown

Inclusive fitness

- Helping others reproduce as they share similar genes
 - o Ie. they are related

- Eg. Male dolphins

Hybrid Zone

- Region where two species overlap and produce hybrids